t(3;11)(q12;p15) NUP98/LNP1

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Abstract

Review on t(3:11)(q12;p15) NUP98/LNP1, with data on clinics, and the genes implicated.

Clinics and pathology

Disease

Myelodysplastic syndrome (MDS), acute myeloid leukemia (AML), and T-cell acute lymphoblastic leukemia (T-ALL)

Phenotype/cell stem origin

Six cases are available: one case of MDS, two cases of AML not otherwise specified, one M2-AML, and two T-ALL cases, one being a case of early T-cell precursor leukaemia (Romana et al., 2006; Chen et al., 2007; Gorello et al., 2008; Coustan-Smith et al., 2009; Lugthart et al., 2010).

Epidemiology

There was 4 male and 2 female patients. Ages were: 3, 28, and 30 years in myeloid cases, and 16 and 36 years in T-ALLs.

Prognosis

Data on prognosis is very scarce: two AML cases died 11 and 23 months after diagnosis, and the early T-cell precursor leukaemia phenotype, in this study of 17 cases with various karyotypes, was said to bear a poor prognosis, but no individual data is available (Coustan-Smith et al., 2009).

Cytogenetics

Cytogenetics morphological

The t(3:11)(q12;p15) was the sole anomaly in four of six cases. +19 was found in one T-ALL.

Genes involved and proteins

NUP98

Location

11p15.4

Protein

Component of nuclear pore complex. NUP98 is found in the nucleoplasmic and cytoplasmic sides of the nuclear pore complex, and functions as nuclear import and nuclear export mRNA factor. NUP98 has a role in the regulation of gene expression via EP300. NUP98 appears to be involved in mitotic spindle formation and cell cycle progression (review in Iwamoto et al., 2010).

LNP1

Location

3q12.2

Note

Also named NP3 or LOC348801.

DNA/RNA

Four exons, the first exon is non-coding.

Protein

Protein of unknown function. 178 amino acids, 21 kDa.

NUP98 was found fused to LNP1 in cases with molecular studies (Romana et al., 2006; Gorello et al., 2008).
Result of the chromosomal anomaly

Hybrid gene

Description
Nucleotide 1718 (exon 13) of NUP98 was fused in-frame with nucleotide 1248 (exon 2) of LNP1. The reciprocal LNP1-NUP98 fusion transcript was also present (Gorello et al., 2008).

Fusion protein

Description
The protein fuses the NUP98 FG repeat motifs and GLEBS-like motif to the entire LNP1, at the start of LNP1 exon 2 (Gorello et al., 2008).

References


This article should be referenced as such: